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L3	L2 and (mutat$ or delet$ or polymorph$)	12	L3
L2	SPG4 or spastin	19	L2
L1	autosomal dominant hereditary spastic paraplegia or AD-HSP	1	L1

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L1 110 AUTOSOMAL DOMINANT HEREDITARY SPASTIC PARAPLEGIA OR AD-HSP

=> s l1 and (spg4 or spastin)
L2 68 L1 AND (SPG4 OR SPASTIN)

=> s (spg4 or spastin) (3a) (mutat? or delet? or polymorph?)
L3 110 (SPG4 OR SPASTIN) (3A) (MUTAT? OR DELET? OR POLYMORPH?)

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L5 ANSWER 1 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 1
AN 2003:317619 BIOSIS
DN PREV200300317619
TI Neurophysiological findings in SPG4 patients differ from other types of spastic paraplegia.
AU Schulte, T.; Miterski, B.; Boernke, C.; Przuntek, H.; Epplen, J. T.; Schoels, L. [Reprint Author]
CS Department of Neurology, St. Josef Hospital, Ruhr-University Bochum, Gudrunstr. 56, D-44791, Bochum, Germany
Ludger Schoels@ruhr-uni-bochum.de
SO Neurology, (May 13, 2003) Vol. 60, No. 9, pp. 1529-1532. print.
ISSN: 0028-3878 (ISSN print)
DT Article
LA English
ED Entered STN: 9 Jul 2003
Last Updated on STN: 9 Jul 2003
AB The authors examined 12 families with ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** for phenotypic characteristics predicting the underlying genotype. They found no clinical differences between patients with or without ***mutations*** in the ***spastin*** gene (SPG4). Motor evoked potentials and nerve conduction studies were almost normal in those with SPG4. In contrast, non-SPG4 families had prolonged central motor conduction times or marked peripheral neuropathy, or both.

L5 ANSWER 2 OF 24 CAPLUS COPYRIGHT 2003 ACS on STN
AN 2003:136931 CAPLUS
DN 138:335737
TI Screening of patients with hereditary spastic paraplegia reveals seven novel ***mutations*** in the ***SPG4*** (***spastin***) gene
AU Proukakis, C.; Auer-Grumbach, M.; Wagner, K.; Wilkinson, P. A.; Reid, E.; Patton, M. A.; Warner, T. T.; Crosby, A. H.
CS Department of Medical Genetics, St. George's Hospital Medical School, University of London, London, SW17 0RE, UK
SO Human Mutation (2003), 21(2), 579/1-579/5
CODEN: HUMUE3, ISSN: 1059-7794
PB Wiley-Liss, Inc.
DT Journal
LA English
AB Hereditary spastic paraplegia (HSP) is a heterogeneous condition characterized in its pure form by progressive lower limb spasticity. ***Mutations*** in ***SPG4*** (encoding ***spastin***) may be responsible for up to 40% of autosomal dominant (AD) cases. A cohort of 41 mostly pure HSP patients from Britain and Austria, 30 of whom displayed AD inheritance, was screened for ***mutations*** in ***SPG4*** by single strand conformation polymorphism (SSCP) anal. followed by sequencing of samples with mobility shifts. The authors identified eight ***SPG4*** ***mutations*** in pure ***AD*** ***HSP*** patients, seven of which were novel: one missense mutation within the AAA cassette (1633G>T), two splice site mutations (1130-1G>T, 1853+2T>A) and four frameshift mutations (190_208dup19, 1259_1260delGT, 1702_1705delGAAG, 1845delG). A novel duplication in intron 11 (1538+42_45dupTATA) was also detected. The authors report the HUGO-approved nomenclature of these mutations as well. Furthermore, the authors detected a silent change (1004G>A, P293P), previously reported as a mutation, which was also present in controls. The frequency of ***SPG4*** ***mutations*** detected in pure ***AD*** ***HSP*** was 33.3%, suggesting that screening of such patients for ***SPG4*** ***mutations*** is worthwhile. Most patients will have unique mutations. Screening of SPG4 in apparently isolated cases of HSP may be of less value.
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L5 ANSWER 3 OF 24 EMBASE COPYRIGHT 2003 ELSEVIER INC. ALL RIGHTS RESERVED
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AN 2003360869 EMBASE
TI Identification of the Drosophila melanogaster homolog of the human spastin gene
AU Kammermeier L.; Spring J.; Stierwald M.; Burgunder J.-M.; Reichert H.
CS L. Kammermeier, Institute of Zoology, Biozentrum and Pharmazentrum, University of Basel, Klingelbergstrasse 50, 4056 Basel, Switzerland.
Lars.Kammermeier@unibas.ch
SO Development Genes and Evolution, (1 Aug 2003) 213/8 (412-415)
Refs: 12
ISSN: 0949-944X CODEN: DGEVFT
CY Germany
DT Journal; Article
FS 005 General Pathology and Pathological Anatomy
008 Neurology and Neurosurgery
021 Developmental Biology and Teratology
022 Human Genetics
LA English
SL English
AB The human SPG4 locus encodes the spastin gene, which is responsible for the most prevalent form of ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** (***AD*** - ***HSP***), a neurodegenerative disorder. Here we identify the predicted gene product CG5977 as the Drosophila homolog of the human spastin gene,

with much higher sequence similarities than any other related AAA domain protein in the fly Furthermore we report a new potential transmembrane domain in the N-terminus of the two homologous proteins During embryogenesis, the expression pattern of Drosophila spastin becomes restricted primarily to the central nervous system, in contrast to the ubiquitous expression of the vertebrate spastin genes Given this nervous system-specific expression, it will be important to determine if *Drosophila* ***spastin*** loss-of-function ***mutations*** also lead to neurodegeneration.

L5 ANSWER 4 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC on STN
AN 2003:138570 BIOSIS
DN PREV200300138570
TI Screening of patients with hereditary spastic paraplegia reveals seven novel ***mutations*** in the ***SPG4*** (***Spastin***) gene
AU Proukakis, C.; Auer-Grumbach, M.; Wagner, K.; Wilkinson, P. A.; Reid, E.; Patton, M. A.; Warner, T. T.; Crosby, A. H. [Reprint Author]
CS Medical Genetics, St George's Hospital, Cranmer Terrace, London, SW17 0RE,
UK
acrosby@sghms.ac.uk
SO Human Mutation, (2003) Vol. 21, No. 2, pp. 170 print
ISSN: 1059-7794.
DT Article
LA English
ED Entered STN: 12 Mar 2003
Last Updated on STN: 12 Mar 2003
AB Hereditary spastic paraplegia (HSP) is a heterogeneous condition characterised in its pure form by progressive lower limb spasticity ***Mutations*** in ***SPG4*** (encoding ***spastin***) may be responsible for up to 40% of autosomal dominant (AD) cases A cohort of 41 mostly pure HSP patients from Britain and Austria, 30 of whom displayed AD inheritance, was screened for ***mutations*** in ***SPG4*** by single strand conformation polymorphism (SSCP) analysis followed by sequencing of samples with mobility shifts We identified eight ***SPG4*** ***mutations*** in pure ***AD*** ***HSP*** patients, seven of which were novel one missense mutation within the AAA cassette (1633>T), two splice site mutations (1130-1G>T, 1853+2T>A) and four frameshift mutations (190 - 208dup19, 1259 - 1260delGT, 1702 - 1705delGAAG, 1845delG). A novel duplication in intron 11 (1538+42 - 45dupTATA) was also detected We report the HUGO-approved nomenclature of these mutations as well Furthermore, we detected a silent change (1004G>A; P293P), previously reported as a mutation, which was also present in controls The frequency of ***SPG4*** ***mutations*** detected in pure ***AD*** ***HSP*** was 33.3%, suggesting that screening of such patients for ***SPG4*** ***mutations*** is worthwhile. Most patients will have unique mutations. Screening of SPG4 in apparently isolated cases of HSP may be of less value

L5 ANSWER 5 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC on STN
DUPLICATE 3
AN 2003:51347 BIOSIS
DN PREV200300051347
TI ***Mutations*** of ***SPG4*** are responsible for a loss of function of spastin, an abundant neuronal protein localized in the nucleus.
AU Charvin, Delphine; Cifuentes-Diaz, Carmen; Fonknechten, Nuria; Joshi, Vandana; Hazan, Jamile; Melki, Judith [Reprint Author]; Betuing, Sandrine
CS Molecular Neurogenetics Laboratory, INSERM, Universite d'Evry, E-0223, GENOPOLE, 2 Rue Gaston Cremieux, 91057, CP5724, Evry, France
j melki@genopole inserm fr
SO Human Molecular Genetics, (1 January, 2003) Vol. 12, No. 1, pp. 71-78 print.
ISSN: 0964-6906 (ISSN print)
DT Article
LA English
ED Entered STN: 22 Jan 2003
Last Updated on STN: 22 Jan 2003
AB ***Mutations*** of ***spastin*** are responsible for the most common autosomal dominant form of hereditary spastic paraplegia (***AD*** - ***HSP***), a disease characterized by axonal degeneration of corticospinal tracts and posterior columns Generation of polyclonal antibodies specific to spastin has revealed two isoforms of 75 and 80 kDa in both human and mouse tissues with a tissue-specific variability of the isoform ratio Spastin is an abundant protein in neural tissues and immunolabeling experiments have shown that spastin is expressed in neurons but not in glial cells. These data indicate that axonal degeneration linked to ***spastin*** ***mutations*** is caused by a primary defect of neurons Protein and transcript analyses of patients carrying either nonsense or frameshift ***spastin*** ***mutations*** revealed neither truncated protein nor mutated transcripts, providing evidence that these mutations are responsible for a loss of spastin function Identifying agents able to induce the expression of the non-***mutated*** ***spastin*** allele should represent an attractive therapeutic strategy in this disease.

L5 ANSWER 6 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC on STN
DUPLICATE 4
AN 2003:349459 BIOSIS
DN PREV200300349459
TI A novel insertion ***mutation*** in ***spastin*** gene is the cause of spastic paraplegia in a Chinese family
AU Qin, Wei, Zhang, Tao; Han, Ju, Tang, LiQun, Li, Xingwang, Feng, Guoyin, Liu, Wanqing, He, Lin [Reprint Author]
CS Bio-X Life Science Research Center, Shanghai Jiao Tong University, 1954 Hua Shan Road, Hao Ran Building, P.O Box 501, Shanghai, 200030, China
helin@sjtu.edu.cn
SO Journal of the Neurological Sciences, (June 15, 2003) Vol. 210, No. 1-2, pp. 35-39. print.
CODEN: JNSCAG ISSN: 0022-510X.
DT Article
LA English
ED Entered STN: 30 Jul 2003
Last Updated on STN: 30 Jul 2003
AB A total of eight loci for ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** (ADHSP) has been mapped to chromosome 14q, 2p, 15q, 8q, 10q, 12q, 19q, 2q, respectively, among which the SPG4 gene on chromosome 2p21-22 encoding spastin, an ATPase of the AAA family, accounts for 40-50% of all ADHSP families and is expressed in both adult and fetal tissues In this work, we reveal a novel insertion mutation in exon 11 of the SPG4 gene found in a big Chinese family composed of 47 members, including 20 affected ones, using linkage analysis The mutation was well demonstrated to be the cause of loss of production of the functional protein by pre-termination of translation in AAA cassette region To our knowledge, this is the first report of ***spastin*** ***mutation*** in China

L5 ANSWER 7 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC. on STN
AN 2003:110901 BIOSIS
DN PREV200300110901
TI SPG3A An additional family carrying a new atlastin mutation
AU Tessa, A.; Casali, C.; Damiano, M.; Bruno, C.; Fortini, D.; Patrono, C., Cricchi, F.; Valoppi, M.; Nappi, G.; Amabile, G. A.; Bertini, E., Santorelli, F. M. [Reprint Author]
CS Molecular Medicine and Neurology, IRCCS-Bambino Gesu Hospital, Piazza S Onofrio 4, 00165, Rome, Italy
fms3@na.flashnet.it
SO Neurology, (December 24, 2002) Vol. 59, No. 12, pp. 2002-2005 print
ISSN: 0028-3878 (ISSN print)
DT Article
LA English
ED Entered STN: 26 Feb 2003
Last Updated on STN: 26 Feb 2003
AB The authors report on a novel frameshift mutation (c.1688insA) in the SPG3A gene resulting in premature translation termination of the gene product atlastin. These data add a new variant to the second disease gene in ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** (ADHSP) and lend definitive support to its causative role By combining direct testing of SPAST and SPG3A, at least 50% of ADHSP families can now receive appropriate genetic diagnosis

L5 ANSWER 8 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC on STN
DUPLICATE 5
AN 2002:515459 BIOSIS
DN PREV200200515459
TI A novel missense ***mutation*** (I344K) in the ***SPG4*** gene in a Korean family with ***autosomal*** - ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** .
AU Ki, Chang-Seok; Lee, Won Yong, Han, Do Hoon, Sung, Duk Hyun, Lee, Kyung-Bok, Lee, Kyung-A., Cho, Sang Seon; Cho, Seunghee, Hwang, Hyokkee, Sohn, Kwang Min; Choi, Yeun Joo, Kim, Jong-Won [Reprint author]
CS Department of Clinical Pathology, Sungkyunkwan University School of Medicine, Samsung Medical Center, No. 50 Ilwon-Dong, Kangnam-Gu, Seoul, 135-710, South Korea
jwonk@smc.samsung.co.kr
SO Journal of Human Genetics, (2002) Vol. 47, No. 9, pp. 473-477 print
ISSN: 1434-5161.
DT Article
LA English
ED Entered STN: 2 Oct 2002
Last Updated on STN: 2 Oct 2002
AB Hereditary spastic paraplegia (HSP) is a group of clinically and genetically heterogeneous neurodegenerative disorders characterized by slowly progressive spasticity and weakness of the lower extremities. Among eight loci linked with autosomal-dominant (***AD***) ***HSP*** , the SPG4 locus on chromosome 2p22 accounts for about 40% of all patients. Recently, mutations in a new member of the AAA protein family, called spastin, have been identified as responsible for SPG4-linked ***AD*** - ***HSP*** Here, we describe a novel missense mutation (c.1031T>A, I344K) in exon 7 of the SPG4 gene identified in a Korean family with typical clinical features of pure ***AD*** - ***HSP*** The mutation affects the third amino acid of the highly conserved AAA cassette domain, which is the most fore part of the domain altered by a missense mutation reported so far. Clinical presentations of affected individuals carrying the I344K mutation were not different from those of pure ***AD*** - ***HSP*** with ***SPG4*** ***mutations*** reported previously. However, it is noteworthy that neither urinary dysfunction nor involvement of upper extremities was noticed in this family To our knowledge, this is the first report of genetically confirmed ***AD*** - ***HSP*** in Korea

L5 ANSWER 9 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2002:624816 BIOSIS
DN PREV200200624816
TI *Molecular diagnostic testing for* ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** : Identification of novel ***mutations*** in the ***SPG4*** gene
AU Wang, J. [Reprint author]; Hennigan, A. N. [Reprint author]; Morini, A. [Reprint author]; Ananth, U. [Reprint author]; Seltzer, W. K. [Reprint author]
CS Athena Diagnostics, Inc., Worcester, MA, 01605, USA
SO American Journal of Human Genetics, (October, 2002) Vol. 71, No. 4 Supplement, pp. 386. print.
Meeting Info.: 52nd Annual Meeting of the American Society of Human Genetics. Baltimore, MD, USA. October 15-19, 2002. American Society of Human Genetics.
CODEN: AJHGAG. ISSN: 0002-9297
DT Conference; (Meeting)
Conference, Abstract; (Meeting Abstract)
LA English
ED Entered STN: 12 Dec 2002
Last Updated on STN: 12 Dec 2002

L5 ANSWER 10 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2003:472893 BIOSIS
DN PREV200300472893
TI ***Spastin*** , the protein ***mutated*** in ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** , is involved in microtubule dynamics.
AU Errico, A. [Reprint Author]; Claudiani, P. [Reprint Author]; Ballabio, A. [Reprint Author]; Rugarli, E. I. [Reprint Author]
CS Telethon Institute of Genetics and Medicine, Napoli, Italy
SO European Journal of Human Genetics, (2002) Vol. 10, No. Supplement 1, pp. 262-263. *print.*
Meeting Info.: European Human Genetics Conference 2002 in conjunction with the European Meeting on Psychosocial Aspects of Genetics 2002. Strasbourg, France. May 25-28, 2002. European Society of Human Genetics (ESHG).
ISSN: 1018-4813
DT Conference, (Meeting)
Conference; Abstract; (Meeting Abstract)
LA English
ED Entered STN: 15 Oct 2003
Last Updated on STN: 15 Oct 2003

L5 ANSWER 11 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 6
AN 2002:238084 BIOSIS
DN PREV200200238084
TI Missense and splice site ***mutations*** in ***SPG4*** suggest loss-of-function in dominant spastic paraplegia
AU Patrono, Clance; Casali, Carlo; Tessa, Alessandra; Cricchi, Federica; Fortini, Daniela; Carrozzo, Rosalba; Siciliano, Gabriele; Bertini, Enrico; *Santorelli, Filippo M. [Reprint author]*
CS Molecular Medicine, IRCCS-Children's Hospital Bambino Gesu, Piazza S. Onofrio, 4, 00165, Rome, Italy
fms3@na.flashnet.it
SO Journal of Neurology, (February, 2002) Vol. 249, No. 2, pp. 200-205 print.
CODEN: JNRYA9. ISSN: 0340-5354.
DT Article
LA English
ED Entered STN: 10 Apr 2002
Last Updated on STN: 10 Apr 2002
AB We studied nine Italian families with a pure form of autosomal dominant spastic paraplegia (ADHSP) to assess the frequency of ***mutations*** in the ***SPG4*** gene. We observed marked intrafamilial variability in both age-at-onset and clinical severity, ranging from severe congenital presentation to mild involvement after age 55 years to healthy carriers of the mutation after age 70. Four of nine probands harboured ***SPG4*** ***mutations*** . We identified three new ***SPG4*** ***mutations*** , all predicting a loss-of-function with apparently important consequences for spastin function. RT-PCR studies predict *loss-of-function as a possible mechanism leading to spastin-related HSP.* The current study expands the spectrum of allelic variants in SPG4, confirming their pathological significance in pure ***AD*** - ***HSP*** and suggesting implications for the presumed function of spastin.

L5 ANSWER 12 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 7
AN 2002:175014 BIOSIS
DN *PREV200200175014*
TI ***Spastin*** , the protein ***mutated*** in ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** , is involved in microtubule dynamics
AU Errico, Alessia; Ballabio, Andrea; Rugarli, Elena I. [Reprint author]
CS Telethon Institute of Genetics and Medicine, Via P. Castellino 111, 80131, Naples, Italy
rugarli@tigem.it
SO Human Molecular Genetics, (15 January, 2002) Vol. 11, No. 2, pp. 153-163 print.
ISSN: 0964-6906.
DT Article
LA English
ED Entered STN: 6 Mar 2002
Last Updated on STN: 6 Mar 2002
AB Hereditary spastic paraplegia (HSP) is characterized by progressive weakness and spasticity of the lower limbs, caused by the specific degeneration of the corticospinal tracts, the longest axons in humans. Most cases of the autosomal dominant form of the disease are due to ***mutations*** in the ***SPG4*** gene, which encodes spastin, an ATPase belonging to the AAA family. The cellular pathways in which spastin operates and its role in causing degeneration of motor axons are currently unknown. By expressing wild-type or ATPase-defective spastin in several cell types, we now show that spastin interacts dynamically with microtubules. Spastin association with the microtubule cytoskeleton is mediated by the N-terminal region of the protein, and is regulated through the ATPase activity of the AAA domain. Expression of all the missense mutations into the AAA domain, which were previously identified in patients, leads to constitutive binding to microtubules in transfected cells and induces the disappearance of the aster and the formation of thick perinuclear bundles, suggesting a role of spastin in microtubule dynamics. Consistently, wild-type spastin promotes microtubule disassembly in transfected cells. These data suggest that spastin may be involved in microtubule dynamics similarly to the highly homologous microtubule-severing protein, katanin. Impairment of fine regulation of the microtubule cytoskeleton in long axons, due to ***spastin*** ***mutations*** , may underlie pathogenesis of HSP.

L5 ANSWER 13 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 8
AN 2002:461029 BIOSIS
DN PREV200200461029
TI ***Mutation*** analysis of the ***spastin*** gene (SPG4) in patients in Germany with ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia***
AU Sauter, S. [Reprint author], Miterski, B., Klimpe, S.; Boensch, D.; Schoels, L., Visbeck, A., Papke, T.; Hopf, H. C.; Engel, W.; Deufel, T.; Epplen, J. T., Neesen, J.
CS Institute of Human Genetics, University of Goettingen, Heinrich-Dueker-Weg 12, 37073, Goettingen, Germany
ssauter@gwdg.de
SO Human Mutation, (2002) Vol. 20, No. 2, pp. 127-132. print.
ISSN: 1059-7794.
DT Article
LA English
OS Genbank-AJ246001; EMBL-AJ246001, DDBJ-AJ246001, Genbank-AJ246003; EMBL-AJ246003, DDBJ-AJ246003
ED Entered STN: 28 Aug 2002
Last Updated on STN: 28 Aug 2002
AB Hereditary spastic paraplegias (HSP) comprise a genetically and clinically heterogeneous group of neurodegenerative disorders characterized by *progressive spasticity and hyperreflexia of the lower limbs.* ***Autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** 4 linked to chromosome 2p (SPG4) is the most common form of ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** It is caused by ***mutations*** in the ***SPG4*** gene encoding spastin, a member of the AAA protein family of ATPases. In this study the spastin gene of HSP patients from 161 apparently unrelated families in Germany was analyzed. The authors identified mutations in 27 out of the 161 HSP families, 23 of these mutations have not been described before and only one mutation was found *in two families. Among the detected mutations are* 14 *frameshift*, four nonsense, and four missense mutations, one large deletion spanning several exons, as well as four mutations that affect splicing. Most of the novel mutations are located in the conserved AAA cassette-encoding region of the spastin gene. The relative frequency of ***spastin*** gene ***mutations*** in an unselected group of German HSP patients is approximately 17%. Frameshift mutations account for the majority of ***SPG4*** ***mutations*** in this population. The proportion of splice mutations is considerably lower than reported elsewhere.

L5 ANSWER 14 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 9
AN 2002:525172 BIOSIS
DN PREV200200525172
TI Three novel ***spastin*** (***SPG4***) ***mutations*** in families with ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia***
AU Proukakis, Christos; Hart, Paul E.; Cornish, Amy; Warner, Thomas T.; Crosby, Andrew H. [Reprint author]
CS *Department of Medical Genetics, St George's Hospital Medical School,* Cranmer Terrace, London, SW17 0RE, UK
acrosby@sghms.ac.uk
SO Journal of the Neurological Sciences, (September 15, 2002) Vol. 201, No. 1-2, pp. 65-69. print
CODEN: JNSCAG. ISSN: 0022-510X
DT Article
LA English

ED Entered STN. 9 Oct 2002
Last Updated on STN: 9 Oct 2002
AB Hereditary spastic paraplegia (HSP) is a clinically and genetically heterogeneous condition, characterised principally by progressive spasticity of the lower limbs. Forty percent of autosomal dominant (AD) pedigrees show linkage to the SPG4 locus on chromosome 2, which encodes spastin, an ATPase associated with diverse cellular activities (AAA) protein. We have performed a clinical and genetic study of three ***AD*** - ***HSP*** families linked to SPG4. Sequencing revealed three novel causative mutations. Two of the mutations were located in exon 5 (a 1-base pair (bp) insertion and a 5-bp deletion), resulting in frameshift and premature termination of translation, with the predicted protein lacking the entire AAA functional domain. The 5-bp deletion was associated with a later onset and mild cerebellar features. The third mutation was a 3-bp deletion in exon 9, resulting in the loss of a highly conserved phenylalanine residue within the AAA cassette and an apparently milder phenotype. This is the first example of a deletion of an amino acid in spastin.

L5 ANSWER 15 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 10
AN 2002:378954 BIOSIS
DN PREV200200378954
TI A novel ***mutation*** in the ***spastin*** gene in a family with spastic paraplegia
AU Morita, Mitsuya [Reprint author]; Ho, Mac; Hosler, Betsy A.; McKenna-Yasek, Diane; Brown, Robert H., Jr.
CS Department of Neurology, Jichi Medical School, 3311-1 Yakushiji, Minamikawachi-machi, Tochigi, 329-0498, Japan
morita-jci@umin.ac.jp
SO Neuroscience Letters, (May 31, 2002) Vol. 325, No. 1, pp. 57-61. print.
CODEN: NELED5. ISSN: 0304-3940
DT Article
LA English
ED Entered STN: 10 Jul 2002
Last Updated on STN: 10 Jul 2002
AB Hereditary spastic paraplegia (HSP) is a degenerative neuromuscular disease characterized by progressive lower extremity weakness, spasticity and hyperreflexia. Inheritance of HSP is commonly autosomal dominant, spastin was identified as the defective gene in chromosome 2p-linked ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** (***AD*** - ***HSP***). In a large American family with ***AD*** - ***HSP*** , we have identified a novel ***spastin*** ***mutation*** at a splice-acceptor site in intron 6 (1130-1 g fwdarw a) and detected a corresponding aberrant transcript generated from a cryptic splice site. This is predicted to cause a frameshift and premature truncation of the abnormal spastin protein. Our data are the first to confirm that a mutation in an acceptor site in the spastin gene results in activation of a cryptic acceptor site and a translational frameshift. The clinical phenotype of this pedigree is also discussed.

L5 ANSWER 16 OF 24 CAPLUS COPYRIGHT 2003 ACS on STN
AN 2002:17039 CAPLUS
DN 136:399839
TI A second leaky splice-site ***mutation*** in the ***spastin*** gene
AU Svenson, Ingrid K.; Ashley-Koch, Allison E.; Pericak-Vance, Margaret A.; Marchuk, Douglas A.
CS Department of Genetics, Duke University Medical Center, Durham, NC, USA
SO American Journal of Human Genetics (2001), 69(6), 1407-1409
CODEN: AJHGAG; ISSN: 0002-9297
PB University of Chicago Press
DT Journal
LA English
AB The splice-site mutation and the extent of missplicing caused by this mutation in ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** was studied. This mutation, an IVS11+2t insertion, causes skipping of exon 11, as detd. by reverse transcriptase-polymerase chain reaction anal. of patient-derived RNA. It would also shift the base pairing by one nucleotide, resulting in a net loss of 4 base pairs relative to the pairing with the wild-type sequence. The findings provide an addnl. support for the hypothesis that the function of spastin is highly concn. dependent. Normally spliced transcript is produced from at least 2 different mutant alleles, which is in agreement with the threshold of spastin required for transition from normal function to disease state lying with narrower interval than the 50% decrease predicted by a disease model of haploinsufficiency.
RE.CNT 3 THERE ARE 3 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L5 ANSWER 17 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2001:556316 BIOSIS
DN PREV200100556316
TI Mutations in a newly identified GTPase gene cause ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia***
AU Zhao, Xinping; Alvarado, David; Rainier, Shirley; Lemons, Rosemary; Hedera, Peter; Weber, Christian H.; Tukel, Turgut; Apak, Memnune; Heiman-Patterson, Terry; Ming, Lei; Bui, Melanie; Fink, John K. [Reprint author]
CS Department of Neurology, University of Michigan, Ann Arbor, MI, 48109, USA
jkfink@umich.edu
SO Nature Genetics, (November, 2001) Vol. 29, No. 3, pp. 326-331. print.
ISSN: 1061-4036
DT Article
LA English
OS Genbank-AF131801, Genbank-AY032844
ED Entered STN: 5 Dec 2001
Last Updated on STN: 25 Feb 2002
AB The hereditary spastic paraplegias (HSPs, Strumpell-Lorrain syndrome, MIM number 18260) are a diverse class of disorders characterized by insidiously progressive lower-extremity spastic weakness. Eight autosomal dominant HSP (ADHSP) loci have been identified, the most frequent of which is that linked to the SPG4 locus on chromosome 2p22 (found in ~42%), followed by that linked to the SPG3A locus on chromosome 14q11-q21 (in approx9%). Only SPG4 has been identified as a causative gene in ADHSP. Its protein (spastin) is predicted to participate in the assembly or function of nuclear protein complexes. Here we report the identification of mutations in a newly identified GTPase gene, SPG3A, in ADHSP affected individuals.

L5 ANSWER 18 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 11
AN 2001:277674 BIOSIS
DN PREV200100277674
TI A large Japanese SPG4 family with a novel insertion ***mutation*** of the ***SPG4*** gene. A clinical and genetic study
AU Namekawa, Michito; Takiyama, Yoshihisa [Reprint author]; Sakoe, Kumi; Shimazaki, Haruo; Amaike, Miho; Nijima, Kenji; Nakano, Imaharu; Nishizawa, Masatoyo
CS Department of Neurology, Jichi Medical School, Kawachi, Tochigi, 329-0498, Japan
ytakiya@ms.jichi.ac.jp
SO Journal of the Neurological Sciences, (March 15, 2001) Vol. 185, No. 1, pp. 63-68. print.
CODEN: JNSCAG. ISSN: 0022-510X.
DT Article
LA English
ED Entered STN: 13 Jun 2001
Last Updated on STN: 19 Feb 2002
AB We studied a large Japanese family with autosomal dominant pure hereditary spastic paraplegia (ADPHSP) clinically and genetically. To date, seven loci causing ADPHSP have been mapped to chromosomes 14q, 2p, 15q, 8q, 12q, 2q, and 19q. Among these loci, the SPG4 locus on chromosome 2p21-p22 has been shown to account for approximately 40% of all ***autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** (ADHSP) families. Very recently, Hazan et al. identified the SPG4 gene encoding a new member of the AAA (ATPases associated with diverse cellular activities) protein family, named spastin. We found a novel insertion mutation (nt1272-1273insA) in exon 8 of the SPG4 gene in the present family. Our study is the first to confirm the causative ***mutation*** of the ***SPG4*** gene in Japanese. Clinically, it is noteworthy that the disease progression in the patients of this family was slow in spite of the late onset, and more than half of the patients showed severe constipation in addition to pure spastic paraplegia.

L5 ANSWER 19 OF 24 EMBASE COPYRIGHT 2003 ELSEVIER INC. ALL RIGHTS RESERVED.
on STN
AN 2001041129 EMBASE
TI Phenotype of ***AD*** - ***HSP*** due to mutations in the SPAST gene: Comparison with ***AD*** - ***HSP*** without mutations.
AU McMonagle P.; Byrne P.C.; Fitzgerald B.; Webb S.; Parfrey N.A.; Hutchinson M.
CS Dr. P. McMonagle, Department of Neurology, St. Vincent's University Hospital, Elm Park, Dublin 4, Ireland. p.mcmonagle@st-vincents.ie
SO Neurology, (26 Dec 2000) 55/12 (1794-1800)
Refs: 39
ISSN: 0028-3878 CODEN: NEURAI
CY United States
DT Journal; Article
FS 008 Neurology and Neurosurgery
022 Human Genetics
LA English
SL English
AB Background: "Pure" autosomal dominant hereditary spastic paraparesis (***AD*** - ***HSP***) is clinically and genetically heterogeneous. There are at least seven genetic loci with varying ages at onset and disability. The SPAST gene at the SPG4 locus on chromosome 2p is the major disease gene for ***AD*** - ***HSP*** . Objectives: To investigate whether there are distinct clinical features among families with ***AD*** - ***HSP*** due to SPAST mutations compared with families excluded from SPG4. Methods. Nineteen families with "pure" ***AD*** - ***HSP*** were identified, and the clinical features of family members were compared using a standard protocol. With use of genetic studies, the families were divided into two groups for comparison. Those with mutations in SPAST, the "mutation-positive" group, and those excluded from SPG4 on the basis of linkage studies, the "SPG4-excluded" group. Results: Twenty-nine individuals from four families had mutations in SPAST, whereas 22 individuals from three families comprised the SPG4-excluded group; in

11 families, the pattern of linkage was unknown In the one remaining family, no mutations were found despite strong linkage to ***SPG4*** Different ***mutations*** were identified in the four SPAST pedigrees, but the clinical picture was similar in each Comparison of the mutation positve group with the SPG4-excluded group revealed an older age at onset (p = 0.03), more disability (p = 0.001), more rapidly progressive paraparesis (p = 0 044), and more cognitive impairment (p = 0 024) among affected individuals with SPAST mutations, not confounded by disease duration. Conclusion: Despite different mutations, SPAST families have a similar phenotype that can be distinguished from other genetic groups

L5 ANSWER 20 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC on STN
DUPLICATE 12
AN 2001.65528 BIOSIS
DN PREV200100065528
TI Novel ***mutations*** in ***spastin*** gene and absence of correlation with age at onset of symptoms.
AU Hentati, A., Deng, H.-X.; Zhai, H., Chen, W.; Yang, Y.; Hung, W.-Y.; Azim, A. C.; Bohlega, S.; Tandan, R.; Warner, C., Laing, N. G.; Cambi, F.; Mitsumoto, H., Roos, R. P.; Boustany, R.-M.; Ben Hamida, M., Hentati, F.; Siddique, T. [Reprint author]
CS Northwestern University Medical School, 300 East Superior St., Tarry Building, Room 13-715, Chicago, IL, 60611, USA
t-siddique@nwu.edu
SO Neurology, (November 14, 2000) Vol. 55, No. 9, pp. 1388-1390. print
CODEN: NEURAI. ISSN: 0028-3878.
DT Article
LA English
ED Entered STN: 31 Jan 2001
Last Updated on STN: 12 Feb 2002
AB ***Autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** is genetically heterogeneous, with at least five loci identified by linkage analysis Recently, ***mutations*** in ***spastin*** were identified in SPG4, the most common locus for dominant hereditary spastic paraplegia that was previously mapped to chromosome 2p22 We identified five novel ***mutations*** in the ***spastin*** gene in five families with ***SPG4*** ***mutations*** from North America and Tunisia and showed the absence of correlation between the predicted mutant spastin protein and age at onset of symptoms

L5 ANSWER 21 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC. on STN
DUPLICATE 13
AN 2001.348 BIOSIS
DN PREV200100000348
TI Hereditary spastic paraplegia caused by ***mutations*** in the ***SPG4*** gene
AU Buerger, Joachim [Reprint author]; Fonknechten, Nuria; Hoeltzenbein, Maria, Neumann, Luitgart; Bratanoff, Elfriede; Hazan, Jamile; Reis, Andre
CS Institut fuer Humangenetik, Charite, Augustenburger Platz 1, Campus Virchow-Klinikum, 13353, Berlin, Germany
joachim.buerger@charite.de
SO European Journal of Human Genetics, (October, 2000) Vol. 8, No. 10, pp 771-776. print.
ISSN: 1018-4813
DT Article
LA English
ED Entered STN: 21 Dec 2000
Last Updated on STN: 21 Dec 2000
AB ***Autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** (***AD*** - ***HSP***) is a genetically heterogeneous neurodegenerative disorder characterised by progressive spasticity of the lower limbs The SPG4 locus at 2p21-p22 accounts for 40-50% of all ***AD*** - ***HSP*** families. The SPG4 gene was recently identified. It is ubiquitously expressed in adult and foetal tissues and encodes spastin, an ATPase of the AAA family We have now identified four novel ***SPG4*** ***mutations*** in German ***AD*** - ***HSP*** families, including one large family for which anticipation had been proposed. Mutations include one frame-shift and one missense mutation, both affecting the Walker motif B. Two further mutations affect two donor splice sites in introns 12 and 16, respectively. RT-PCR analysis of both donor splice site mutations revealed exon skipping and reduced stability of aberrantly spliced SPG4 mRNA. All mutations are predicted to cause loss of functional protein. In conclusion, we confirm in German families that ***SPG4*** ***mutations*** cause ***AD*** - ***HSP*** . Our data suggest that ***SPG4*** ***mutations*** exert their dominant effect not by gain of function but by haploinsufficiency. If a threshold level of spastin were critical for axonal preservation, such threshold dosage effects might explain the variable expressivity and incomplete penetrance of SPG4-linked ***AD*** - ***HSP*** .

L5 ANSWER 22 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC. on STN
DUPLICATE 14
AN 2000.190916 BIOSIS
DN PREV200000190916
TI Spectrum of ***SPG4*** ***mutations*** in autosomal dominant spastic paraplegia.
AU Fonknechten, Nuria; Mavel, Delphine; Byrne, Paula; Davoine, Claire-Sophie; Cruaud, Corinne, Boentsch, Dominikus; Samson, Delphine, Coutinho, Paula, Hutchinson, Michael, McMonagle, Paul; Burgunder, Jean-Marc; Tartaglione, Antonio; Heinzlef, Olivier; Feki, Imed; Deufel, Thomas; Parfrey, Nollaig, Brice, Alexis, Fontaine, Bertrand; Prud'homme, Jean-Francois; Weissenbach, Jean, Durr, Alexandra; Hazan, Jamile [Reprint author]
CS Genoscope, 2 Rue Gaston Cremieux, 91000, Evry, France
SO Human Molecular Genetics, (March 1, 2000) Vol. 9, No. 4, pp. 637-644 print.
ISSN: 0964-6906.
DT Article
LA English
ED Entered STN: 17 May 2000
Last Updated on STN: 4 Jan 2002
AB ***Autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** (***AD*** - ***HSP***) is a group of genetically heterogeneous neurodegenerative disorders characterized by progressive spasticity of the lower limbs Five ***AD*** - ***HSP*** loci have been mapped to chromosomes 14q, 2p, 15q, 8q and 12q. The SPG4 locus at 2p21-p22 has been shown to account for apprx40% of all ***AD*** - ***HSP*** families. SPG4 encoding spastin, a putative nuclear AAA protein, has recently been identified. Here, sequence analysis of the 17 exons of SPG4 in 87 unrelated ***AD*** - ***HSP*** patients has resulted in the detection of 34 novel mutations. These ***SPG4*** ***mutations*** are scattered along the coding region of the gene and include all types of DNA modification including missense (28%), nonsense (15%) and splice site point (26.5%) mutations as well as deletions (23%) and insertions (7.5%). The clinical analysis of the 238 mutation carriers revealed a high proportion of both asymptomatic carriers (14/238) and patients unaware of symptoms (45/238), and permitted the redefinition of this frequent form of ***AD*** - ***HSP*** .

L5 ANSWER 23 OF 24 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC on STN
AN 2000 490986 BIOSIS
DN PREV200000491107
TI Spastin, a new AAA protein, binds to alpha and beta tubulins.
AU Azim, A. C [Reprint author]; Hentati, A [Reprint author]; Haque, M. F U [Reprint author], Hirano, M. [Reprint author], Ouachi, K. [Reprint author], Siddique, T. [Reprint author]
CS Neurology, Northwestern Medical School, Chicago, IL, USA
SO American Journal of Human Genetics, (October, 2000) Vol 67, No 4 Supplement 2, pp. 197 print.
Meeting Info.: 50th Annual Meeting of the American Society of Human Genetics. Philadelphia, Pennsylvania, USA October 03-07, 2000. American Society of Human Genetics
CODEN: AJHGAG ISSN: 0002-9297.
DT Conference; (Meeting)
Conference, Abstract; (Meeting Abstract)
LA English
ED Entered STN: 15 Nov 2000
Last Updated on STN: 10 Jan 2002

L5 ANSWER 24 OF 24 EMBASE COPYRIGHT 2003 ELSEVIER INC ALL RIGHTS RESERVED
on STN DUPLICATE 15
AN 1999391382 EMBASE
TI Spastin, a new AAA protein, is altered in the most frequent form of autosomal dominant spastic paraplegia
AU Hazan J.; Fonknechten N.; Mavel D.; Paternotte C.; Samson D.; Artiguenave F.; Davoine C.-S.; Cruaud C.; Durr A.; Wincker P., Brottier P., Cattolico L.; Barbe V.; Burgunder J.-M.; Prud'homme J.-F.; Brice A.; Fontaine B., Heilig R., Weissenbach J.
CS J. Hazan, Genoscope, Evry, France. jamile@genoscope.cns.fr
SO Nature Genetics, (1999) 23/3 (296-303)
Refs: 48
ISSN: 1061-4036 CODEN: NGENEC
CY United States
DT Journal; Article
FS 008 Neurology and Neurosurgery
022 Human Genetics
LA English
SL English
AB ***Autosomal*** ***dominant*** ***hereditary*** ***spastic*** ***paraplegia*** (***AD*** - ***HSP***) is a genetically heterogeneous neurodegenerative disorder characterized by progressive spasticity of the lower limbs Among the four loci causing ***AD*** - ***HSP*** identified so far, the SPG4 locus at chromosome 2p21-p22 has been shown to account for 40-50% of all ***AD*** - ***HSP*** families. Using a positional cloning strategy based on obtaining sequence of the entire SPG4 interval, we identified a candidate gene encoding a new member of the AAA protein family, which we named spastin. Sequence analysis of this gene in seven SPG4-linked pedigrees revealed several DNA modifications, including missense, nonsense and splice-site ***mutations*** . Both ***SPG4*** and its mouse orthologue were shown to be expressed early and ubiquitously in fetal and adult tissues. The sequence homologies and putative subcellular localization of spastin suggest that this ATPase is involved in the assembly or function of nuclear protein complexes

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L1 110 S AUTOSOMAL DOMINANT HEREDITARY SPASTIC PARAPLEGIA OR AD-HSP
L2 68 S L1 AND (SPG4 OR SPASTIN)
L3 110 S (SPG4 OR SPASTIN) (3A) (MUTAT? OR DELET? OR POLYMORPH?)
L4 51 S L1 AND L3
L5 24 DUP REM L4 (27 DUPLICATES REMOVED)

FILE 'STNGUIDE' ENTERED AT 17:43:34 ON 20 OCT 2003

FILE 'BIOSIS, EMBASE, CAPLUS' ENTERED AT 17:56:14 ON 20 OCT 2003

=> s l3 not l4
L6 59 L3 NOT L4

=> dup rem l6
PROCESSING COMPLETED FOR L6
L7 42 DUP REM L6 (17 DUPLICATES REMOVED)

=> s l7 and py<=2000
1 FILES SEARCHED...
L8 9 L7 AND PY<=2000

=> d bib abs 1-
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L8 ANSWER 1 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2001:218484 BIOSIS
DN PREV200100218484
TI Hereditary spastic paraplegias
AU Angelini, C. [Reprint author]; Pegoraro, E. [Reprint author]; Molon, A. [Reprint author]
CS Department of Neurology, University of Padova, Padova, Italy
SO European Journal of Neurology, (November, 2000) Vol. 7, No. Supplement 3, pp. 172. print
Meeting Info.: 5th Congress of the European Federation of Neurological Societies. Copenhagen, Denmark. October 14-18, 2000. European Federation of Neurological Societies.
ISSN: 1351-5101.
DT Conference, (Meeting)
Conference, Abstract; (Meeting Abstract)
LA English
ED Entered STN: 9 May 2001
Last Updated on STN: 18 Feb 2002

L8 ANSWER 2 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2000:513601 BIOSIS
DN PREV200000513601
TI ***Mutation*** analysis of the ***spastin*** gene (SPG4) in patients with hereditary spastic paraparesis.
AU Lindsey, J. C.; Lusher, M. E.; McDermott, C. J.; White, K. D.; Reid, E.; Rubinsztein, D. C.; Bashir, R.; Hazan, J.; Shaw, P. J.; Bushby, K. M. D. [Reprint author]
CS Human Molecular Genetics Unit, School of Biochemistry and Genetics, University of Newcastle upon Tyne, Newcastle upon Tyne, NE2 4AA, UK
SO Journal of Medical Genetics, (October, 2000) Vol. 37, No. 10, pp. 759-765. print.
CODEN: JMDGAE. ISSN: 0022-2593.
DT Article
LA English
ED Entered STN: 29 Nov 2000
Last Updated on STN: 11 Jan 2002
AB Background-Hereditary spastic paraparesis is a genetically heterogeneous condition. Recently, ***mutations*** in the ***spastin*** gene were reported in families linked to the common SPG4 locus on chromosome 2p21-22. Objectives-To study a population of patients with hereditary spastic paraparesis for ***mutations*** in the ***spastin*** gene (SPG4) on chromosome 2p21-22. Methods-DNA from 32 patients (12 from families known to be linked to ***SPG4***) was analysed for ***mutations*** in the ***spastin*** gene by single strand conformational polymorphism analysis and sequencing. All patients were also examined clinically. Results-Thirteen ***SPG4*** ***mutations*** were identified, 11 of which are novel. These mutations include missense, nonsense, frameshift, and splice site mutations, the majority of which affect the AAA cassette. We also describe a nucleotide substitution outside this conserved region which appears to behave as a recessive mutation. Conclusions-Recurrent ***mutations*** in the ***spastin*** gene are uncommon. This reduces the ease of mutation detection as a part of the diagnostic work up of patients with hereditary spastic paraparesis. Our findings have important implications for the presumed function of ***spastin*** and schemes for ***mutation*** detection in HSP patients.

L8 ANSWER 3 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2000:491158 BIOSIS
DN PREV200000491279
TI Five novel ***mutations*** of ***spastin*** gene in chromosome 2-linked autosomal dominant spastic paraplegia (SPG4).
AU Deng, H.-X. [Reprint author]; Zhai, H. [Reprint author]; Chen, W. [Reprint author]; Hung, W.-Y. [Reprint author]; Hentati, A. [Reprint author]; Siddique, T. [Reprint author]
CS Neurology Dept., Northwestern Univ., Chicago, IL, USA
SO American Journal of Human Genetics, (October, 2000) Vol. 67, No. 4 Supplement 2, pp. 372. print.
Meeting Info.: 50th Annual Meeting of the American Society of Human Genetics. Philadelphia, Pennsylvania, USA. October 03-07, 2000. American Society of Human Genetics.
CODEN: AJHGAG. ISSN: 0002-9297.
DT Conference, (Meeting)
Conference, Abstract; (Meeting Abstract)
Conference; (Meeting Poster)
LA English
ED Entered STN: 15 Nov 2000
Last Updated on STN: 10 Jan 2002

L8 ANSWER 4 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2000:488739 BIOSIS
DN PREV200000488860
TI ***Mutation*** analysis of the ***spastin*** gene in hereditary spastic paraplegia type 4. Evidence of aberrant transcript splicing caused by mutations in noncanonical splice site sequences.
AU Svenson, I. K. [Reprint author]; Ashley-Koch, A. E. [Reprint author]; Gaskell, P. C. [Reprint author]; Riney, T. J. [Reprint author]; Warner, C.; Farrell, C. D.; Boustany, R.-M. N. [Reprint author]; Haines, J. L.; Nance, M. A.; Pericak-Vance, M. A. [Reprint author]; Marchuk, D. A. [Reprint author]
CS Duke University Medical Center, Durham, NC, USA
SO American Journal of Human Genetics, (October, 2000) Vol. 67, No. 4 Supplement 2, pp. 375. print.
Meeting Info.: 50th Annual Meeting of the American Society of Human Genetics. Philadelphia, Pennsylvania, USA. October 03-07, 2000. American Society of Human Genetics.
CODEN: AJHGAG. ISSN: 0002-9297.
DT Conference; (Meeting)
Conference, Abstract; (Meeting Abstract)
Conference, (Meeting Poster)
LA English
ED Entered STN: 15 Nov 2000
Last Updated on STN: 10 Jan 2002

L8 ANSWER 5 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2000:488733 BIOSIS
DN PREV200000488854
TI Hereditary spastic paraplegia caused by ***mutations*** in the ***SPG4*** gene.
AU Burger, J. J. [Reprint author]; Fonknechten, N.; Hoeltzenbein, M.; Neumann, L. [Reprint author]; Hazan, J.; Reis, A. [Reprint author]
CS Charite Human Genetics, Humboldt Univ., Berlin, Germany

SO American Journal of Human Genetics, (October, 2000) Vol 67, No 4 Supplement 2, pp 372 print.
Meeting Info.: 50th Annual Meeting of the American Society of Human Genetics Philadelphia, Pennsylvania, USA October 03-07, 2000 American Society of Human Genetics.
CODEN: AJHGAG ISSN: 0002-9297.
DT Conference, (Meeting)
Conference; Abstract, (Meeting Abstract)
Conference; (Meeting Poster)
LA English
ED Entered STN 15 Nov 2000
Last Updated on STN: 10 Jan 2002

L8 ANSWER 6 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN
AN 2000:434238 BIOSIS
DN PREV200000434238
TI Intrafamilial variability in hereditary spastic paraplegia associated with an ***SPG4*** gene ***mutation***
AU Santorelli, F. M. [Reprint author], Patrono, C., Fortini, D., Tessa, A., Comanducci, G.; Bertini, E., Pierallini, A., Amabile, G. A.; Casali, C.
CS Molecular Medicine and Neurology, Ospedale "Bambino Gesu," IRCCS, Piazza
S. Onofrio 4, 00165, Rome, Italy
SO Neurology, (September 12, 2000) Vol 55, No. 5, pp. 702-705 print.
CODEN: NEURAI ISSN: 0028-3878.
DT Article
LA English
ED Entered STN: 11 Oct 2000
Last Updated on STN: 10 Jan 2002
AB The authors studied a family with pure autosomal dominant spastic paraplegia (ADHSP) that showed a marked intrafamilial variability in both age at onset and clinical severity, ranging from severe congenital presentation to mild involvement after age 55. They found a novel ***mutation*** in the ***SPG4*** gene, which segregates with the disease in six patients. The mutation affects the consensus donor splice site of SPG4 intron 16, resulting in a premature termination codon at amino acid 578. The data confirm the pathologic significance of ***SPG4*** ***mutations*** in pure ADHSP and add to the list of known SPG4 allelic variants.

L8 ANSWER 7 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2000:361049 BIOSIS
DN PREV200000361049
TI Molecular analysis of the *SPG4* gene in *Portuguese* families with spastic paraplegia.
AU Ferreirinha, Fatima [Reprint author], Alonso, I. [Reprint author], Vale, J.; Barros, J., Coutinho, P.; Silveira, I. [Reprint author], Sequeiros, J. [Reprint author]
CS UniGENe-IBMC, Porto, Portugal
SO European Journal of Human Genetics, (June, 2000) Vol. 8, No. Supplement 1, pp. 146. print.
Meeting Info.: European Human Genetics Conference 2000. Amsterdam, Netherlands. May 27-February 30, 2000. European Society of Human Genetics ISSN: 1018-4813.
DT Conference, (Meeting)
Conference; Abstract, (Meeting Abstract)
Conference, (Meeting Poster)
LA English
ED Entered STN: 23 Aug 2000
Last Updated on STN: 8 Jan 2002

L8 ANSWER 8 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN
AN 2000:350945 BIOSIS
DN PREV200000350945
TI Clinical and pathologic findings in hereditary spastic paraparesis with ***spastin*** ***mutation***
AU White, K. D.; Ince, P. G.; Lusher, M., Lindsey, J.; Cookson, M., Bashir, R.; Shaw, P. J.; Bushby, K. M. D. [Reprint author]
CS Department of Human Genetics, 19/20 Claremont Place, Newcastle upon Tyne,
NE2 4AA, UK
SO Neurology, (July 12, 2000) Vol. 55, No. 1, pp. 89-94 print.
CODEN: NEURAI. ISSN: 0028-3878
DT Article
LA English
ED *Entered STN: 16 Aug 2000*
Last Updated on STN: 8 Jan 2002
AB Objective: To describe a family with chromosome 2p-linked hereditary spastic paraparesis (HSP) associated with dementia and illustrate the cerebral pathology associated with this disorder. Background: HSP comprises a heterogeneous group of inherited disorders in which the main clinical feature is severe, progressive lower limb spasticity. Nongenetic classification relies on characteristics such as mode of inheritance, age at onset, and the presence or absence of additional neurologic features. Several loci have been identified for autosomal dominant pure HSP. The most common form, which links to chromosome 2p (SPG4), has recently been shown to be due to ***mutations*** in ***spastin***, the gene encoding a novel AAA-containing protein. Results: The authors report four generations of a British family with autosomal dominant HSP in whom haplotype analysis indicates linkage to chromosome 2p. In addition, a missense mutation has been identified in exon 10 of the spastin gene (A1395G). Dementia was documented clinically in one member of the family, two other affected family members were reported to have had late onset memory loss, and a younger affected individual showed evidence of memory disturbance and learning difficulties. Autopsy of the demented patient *confirmed changes in the spinal cord typical of HSP and also demonstrated* specific cortical pathology. There was neuronal depletion and tau-immunoreactive neurofibrillary tangles in the hippocampus and tau-immunoreactive balloon cells were seen in the limbic and neocortex. The substantia nigra showed Lewy body formation. The pathologic findings are not typical of known tauopathies. Conclusions: The authors confirm that chromosome 2p-linked HSP can be associated with dementia and that this phenotype may be associated with a specific and unusual cortical pathology.

L8 ANSWER 9 OF 9 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 2000:277076 BIOSIS
DN PREV200000277076
TI Phenotype of ***SPG4*** ***mutations*** in autosomal dominant hereditary spastic paraparesis
AU McMonagle, Paul [Reprint author]; Byrne, Paula [Reprint author]; Fitzgerald, Brendan [Reprint author], Stewart, Webb [Reprint author]; Parfrey, Nollaig [Reprint author]; Hutchinson, Michael [Reprint author]
CS Dublin, Ireland
SO Neurology, (April 11, 2000) Vol. 54, No. 7 Supp. 3, pp. A424-A425 print
Meeting Info.: 52nd Annual Meeting of the American Academy of Neurology. San Diego, CA, USA April 29-May 06, 2000. American Academy of Neurology
CODEN: NEURAI ISSN: 0028-3878.
DT Conference, (Meeting)
Conference; Abstract, (Meeting Abstract)
LA English
ED Entered STN: 6 Jul 2000
Last Updated on STN: 7 Jan 2002

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FULL ESTIMATED COST		20.64	100.06

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CA SUBSCRIBER PRICE		0.00	-1.30

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FULL ESTIMATED COST		0.18	100.24

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CA SUBSCRIBER PRICE		0.00	-1.30

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Truncation
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MACINTOSH VERSION IS V6.0b(ENG) AND V6.0Jb(JP),
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COST IN U.S. DOLLARS SINCE FILE TOTAL
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=> s SPG4 or spastin
L1 219 SPG4 OR SPASTIN

=> s l1 and py<=1999
1 FILES SEARCHED...
L2 44 L1 AND PY<=1999

=> dup rem l2
PROCESSING COMPLETED FOR L2
L3 23 DUP REM L2 (21 DUPLICATES REMOVED)

=> d bib abs 1-
YOU HAVE REQUESTED DATA FROM 23 ANSWERS - CONTINUE? Y/(N):y

L3 ANSWER 1 OF 23 CAPLUS COPYRIGHT 2003 ACS on STN
AN 1999:811376 CAPLUS
DN 132:45827
TI YAC fragmentation vectors using short triplet repeats as the target sequence for homologous recombination and their uses in phys. mapping human genome
IN Del-Favero, Jurgen; Van Broeckhoven, Christine
PA Vlaams Interuniversitair Instituut Voor Biotechnologie VZW, Belg
SO PCT Int. Appl., 52 pp.
CODEN: PIXXD2
DT Patent
LA English
FAN CNT 1

PATENT NO.	KIND	DATE	APPLICATION NO.	DATE
PI WO 9966059	A1	19991223	WO 1999-EP4106	19990611 <--

W: AL, AM, AT, AU, AZ, BA, BB, BG, BR, BY, CA, CH, CN, CU, CZ, DE, DK, EE, ES, FI, GB, GD, GE, GH, GM, HR, HU, ID, IL, IN, IS, JP, KE, KG, KP, KR, KZ, LC, LK, LR, LS, LT, LU, LV, MD, MG, MK, MN, MW, MX, NO, NZ, PL, PT, RO, RU, SD, SE, SG, SI, SK, SL, TJ, TM, TR, TT, UA, UG, US, UZ, VN, YU, ZW, AM, AZ, BY, KG, KZ, MD, RU, TJ, TM
RW: GH, GM, KE, LS, MW, SD, SL, SZ, UG, ZW, AT, BE, CH, CY, DE, DK, ES, FI, FR, GB, GR, IE, IT, LU, MC, NL, PT, SE, BF, BJ, CF, CG, CI, CM, GA, GN, GW, ML, MR, NE, SN, TD, TG

AU 9945131	A1	20000105	AU 1999-45131	19990611
PRAI EP 1998-201976		19980612		
WO 1999-EP4106		19990611		

AB Novel vectors for liberation of subsequences from yeast artificial chromosomes (YACs), called fragmentation vectors, use short triplet repeats as the target sequence for homologous recombination to ext. sequences from the larger clone. These vectors can be used in large-scale mapping and sequencing projects. The new vectors have one telomere, a selectable marker (Lys2) and one short triplet repeats as the target sequence for homologous recombination, either with or without a centromere. These vectors allow direct acentric and centric fragmentation of yeast artificial chromosomes (YACs) and selection of fragmented YACs contg. triplet repeats sequence in yeast strain AB1380. High recombination efficiencies were obtained in fragmentations of YAC clones contg. SCA7 (spinocerebellar ataxia type 7) gene or ***SPG4*** locus (one of loci for dominant spastic paraplegia) using vectors with a low-copy no. of CAG or CTG triplet repeats. (SCA7 is the causative agent for autosomal dominant cerebellar ataxia with retinal degeneration if 10 of CAG repeats in its exon I expanded to 38). Several sets of fragmented clones were obtained according to their final sizes and all clones with the same size represented a sequence-specific recombination event. Two vectors with a short sequence of CGG or CCG repeats were shown to have even higher recombination efficiency than those with CAG or CTG repeats. These repeats-based fragmentation vectors are esp. useful to discover the abnormality in the polymorphism of short triplet repeats in the flanking regions of specific human genes which might play a role in its aberrant expression and assocd. disorders.
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L3 ANSWER 2 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN
DUPLICATE 1
AN 2000:19189 BIOSIS
DN PREV200000019189
TI Autosomal dominant spastic paraplegia: Refined SPG8 locus and additional genetic heterogeneity.
AU Reid, E.; Dearlove, A. M.; Whiteford, M. L.; Rhodes, M.; Rubinsztein, D. C. [Reprint author]
CS Department of Medical Genetics, Cambridge Institute for Medical Research, Addenbrooke's Hospital, Hills Road, Floor 4, Wellcome/MRC Building, Cambridge, CB2 2XY, UK
SO Neurology, (Nov. 10, 1999) Vol. 53, No. 8, pp. 1844-1849. print.
CODEN: NEURAI. ISSN: 0028-3878.
DT Article
LA English
ED Entered STN: 29 Dec 1999
Last Updated on STN: 31 Dec 2001
AB Objective: To map the gene responsible for autosomal dominant pure hereditary spastic paraplegia (ADPHSP) in a large affected family. Background: Autosomal dominant pure hereditary spastic paraplegia (ADPHSP) is genetically heterogeneous, and loci have been mapped at chromosomes 2p (***SPG4***), 14q (SPG3), 15q (SPG6), and recently, in a single family, at chromosome 8q24 (SPG8). Methods. The authors carried out a genomewide linkage screen on a large family with ADPHSP, for which linkage to the chromosome 2, 14, and 15 loci was excluded. Results. Analysis of markers on chromosome 8q24 gave a peak two-point lod score of 4.49 at marker D8S1799. Analysis of recombination events in this family and in the previously published SPG8-linked family narrowed the SPG8 locus from 6.2 cM to a 3.4-cM region between markers D8S1804 and D8S1179. In another four families, linkage to all four known ADPHSP loci was excluded. The SPG8-linked family had a significantly older mean age at onset of symptoms and had significantly more wheelchair-using patients than the four linkage-excluded families. Conclusions. These results contain the presence of an autosomal dominant pure hereditary spastic paraplegia (ADPHSP) locus at chromosome 8q24 and strongly suggest that there are at least five ADPHSP loci. The data provide additional evidence for locus-phenotype correlations in ADPHSP.

L3 ANSWER 3 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN
AN 1999:520160 BIOSIS
DN PREV199900520160
TI ***SPG4*** : A recombination event narrows the minimum candidate region
AU Svenson, I. K. [Reprint author]; Nance, M. A.; Haines, J. L.; Scott, W. K. [Reprint author]; Pericak-Vance, M. A. [Reprint author]; Marchuk, D. A. [Reprint author]
CS Duke University Medical Center, Durham, NC, USA

SO American Journal of Human Genetics, (Oct., 1999) Vol 65, No. 4, pp. A420. print.
Meeting Info.: 49th Annual Meeting of the American Society of Human Genetics. San Francisco, California, USA October 19-23, 1999 The American Society of Human Genetics
CODEN: AJHGAG ISSN: 0002-9297
DT Conference, (Meeting)
Conference, Abstract, (Meeting Abstract)
Conference; (Meeting Poster)
LA English
ED Entered STN. 3 Dec 1999
Last Updated on STN. 3 Dec 1999

L3 ANSWER 4 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC on STN
DUPLICATE 2
AN 1999:509702 BIOSIS
DN PREV199900509702
TI A fine integrated map of the ***SPG4*** locus excludes an expanded CAG repeat in chromosome 2p-linked autosomal dominant spastic paraplegia
AU Hazan, Jamile [Reprint author], Davoine, Claire-Sophie; Mavel, Delphine, Fonknechten, Nuria, Paternotte, Caroline; Fizames, Cecile, Cruaud, Corinne; Samson, Delphine, Muselet, Delphine; Vega-Czarny, Nathalie, Brice, Alexis; Gyapay, Gabor, Heilig, Roland; Fontaine, Bertrand, Weissenbach, Jean
CS Genoscope, 2 rue Gaston Cremieux, 91000, Evry, France
SO Genomics, (Sept. 15, 1999) Vol 60, No. 3, pp. 309-319. print.
CODEN: GNMCEP ISSN: 0888-7543
DT Article
LA English
ED Entered STN. 3 Dec 1999
Last Updated on STN: 3 Dec 1999
AB Autosomal dominant hereditary spastic paraplegia (AD-HSP) is a genetically heterogeneous disorder characterzed by progressive spasticity of the lower limbs. A major locus (***SPG4***) causing AD-HSP in about 40% of the families was mapped to chromosome 2p. The analysis of six ***SPG4*** -linked AD-HSP families using the RED procedure previously showed the expansion of a CAG repeat in affected individuals. To identify the gene responsible for this form of HSP, we have constructed a 3.5-Mb YAC contig flanked by loci D2S400 and D2S367, have subcloned five of these YACs spanning the candidate region into cosmids, and screened these cosmid libraries for the presence of CAG repeat sequences. Four CAG repeats have been identified but none of them is expanded in 26 patients from 13 ***SPG4*** -linked AD-HSP families. A gene map comprising 21 transcripts was established using expressed sequence tags (ESTs) assigned previously to this region of 2p21-p22 with radiation hybrid panels GeneBridge 4 and G3. Full-lengthcDNAs corresponding to the 14 ESTs mapping to the ***SPG4*** interval flanked by loci D2S352 and D2S2347 were isolated and sequenced. None contains a CAG repeat in its coding sequence. Finally, we have assembled a BAC contig composed of 37 clones that were also screened for the presence of CAG repeats; this failed to detect additional repeats to those identified on YACs

L3 ANSWER 5 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC. on STN
DUPLICATE 3
AN 2000:14627 BIOSIS
DN PREV200000014627
TI ***Spastin*** , a new AAA protein, is altered in the most frequent form of autosomal dominant spastic paraplegia.
AU Hazan, Jamile [Reprint author], Fonknechten, Nuria, Mavel, Delphine, Paternotte, Caroline, Samson, Delphine; Artiguenave, Francois; Davoine, Claire-Sophie; Cruaud, Corinne, Durr, Alexandra; Wincker, Patrick; Brottier, Philippe; Cattolico, Laurence; Barbe, Valerie, Burgunder, Jean-Marc; Prud'homme, Jean-Francois, Brice, Alexis, Fontaine, Bertrand, Heilig, Roland; Weissenbach, Jean
CS Genoscope, Evry, France
SO Nature Genetics, (Nov , 1999) Vol. 23, No. 3, pp 296-303 print
ISSN. 1061-4036.
DT Article
LA English
ED Entered STN. 29 Dec 1999
Last Updated on STN: 31 Dec 2001
AB Autosomal dominant hereditary spastic paraplegia (AD-HSP) is a genetically heterogeneous neurodegenerative disorder characterized by progressive spasticity of the lower limbs. Among the four loci causing AD-HSP identified so far, the ***SPG4*** locus at chromosome 2p21-p22 has been shown to account for 40-50% of all AD-HSP families. Using a positional cloning strategy based on obtaining sequence of the entire ***SPG4*** interval, we identified a candidate gene encoding a new member of the AAA protein family, which we named ***spastin***. Sequence analysis of this gene in seven ***SPG4*** -linked pedigrees revealed several DNA modifications, including missense, nonsense and splice-site mutations. Both ***SPG4*** and its mouse orthologue were shown to be expressed early and ubiquitously in fetal and adult tissues. The sequence homologies and putative subcellular localization of ***spastin*** suggest that this ATPase is involved in the assembly or function of nuclear protein complexes

L3 ANSWER 6 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC. on STN
DUPLICATE 4
AN 2000:6387 BIOSIS
DN PREV200000006387
TI Isolation of CAG/CTG repeats from within the chromosome 2p21-p24 locus for autosomal dominant spastic paraplegia (***SPG4***) by YAC fragmentation.
AU Del-Favero, Jurgen [Reprint author], Goossens, Dirk, De Jonghe, Peter, Benson, Kathleen, Michalik, Andrej, Van den Bossche, Dirk; Horwitz, Marshall; Van Broeckhoven, Christine
CS Psychiatric Genetics Group, Department of Biochemistry, University of Antwerp (UIA), Universiteitsplein 1, B-2610, Antwerp, Belgium
SO Human Genetics, (Sept , 1999) Vol 105, No 3, pp 217-225 print.
CODEN. HUGEDQ ISSN: 0340-6717.
DT Article
LA English
ED Entered STN: 23 Dec 1999
Last Updated on STN. 31 Dec 2001
AB Pure autosomal dominant spastic paraplegia (SPG) is a genetically heterogeneous neurodegenerative disorder of the central nervous system clinically characterized by progressive spasticity mainly affecting the lower limbs. Three distinct loci have been mapped to chromosomes 14q (SPG3), 2p (***SPG4***) and 15q (SPG6). In particular, ***SPG4*** families show striking intrafamilial variability suggestive of anticipation and evidence has been provided that CAG/CTG repeat expansions may be involved. To isolate CAG/CTG repeat containing sequences from within the ***SPG4*** candidate region, a novel approach was developed. Fragmentation vectors were assembled allowing direct fragmentation of yeast artificial chromosomes (YACs) with a short (gtoreq21 bp) CAG/CTG sequence as the target site for homologous recombination. We used the CAG/CTG YAC fragmentation vectors to isolate CAG/CTG containing sequences from four YACs spanning the ***SPG4*** candidate region between D2S400 and D2S367. A total of four CAG/CTG containing sequences were isolated of which three were novel. However, none of the four CAG/CTG repeats showed expanded alleles in two Belgian ***SPG4*** families. In addition, we showed that the CAG/CTG alleles detected by the repeat expansion detection (RED) method could be fully explained by two polymorphic nonpathogenic CAG/CTG repeats on chromosomes 17 and 18, respectively. Also, the RED expansions in six SPG families could not be explained by amplification of the CAG/CTG repeats at the ***SPG4*** locus. Together, our data do not support the hypothesis of a CAG/CTG repeat expansion as the molecular mechanism underlying ***SPG4*** pathology.

L3 ANSWER 7 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC. on STN
DUPLICATE 5
AN 1998:496172 BIOSIS
DN PREV199800496172
TI CAG repeat expansion in autosomal dominant familial spastic paraparesis: Novel expansion in a subset of patients
AU Benson, Kathleen F., Horwitz, Mashall [Reprint author]; Wolff, John, Friend, Kathy, Thompson, Elizabeth, White, Sue, Richards, Robert I ; Raskind, Wendy H , Bird, Thomas D.
CS Markey Mol. Med. Cent., Dep. Med , Sch Med , Univ Wash , 1705 N.E. Pacific St., Box 357720, Seattle, WA 98195-7720, USA
SO Human Molecular Genetics, (Oct., 1998) Vol 7, No 11, pp. 1779-1786. print
ISSN. 0964-6906
DT Article
LA English
ED Entered STN 18 Nov 1998
Last Updated on STN. 18 Nov 1998
AB Autosomal dominant familial spastic paraplegia (FSP) is a genetically heterogeneous neurodegenerative disorder displaying anticipation for which three loci have been mapped to the chromosomal positions 14q11.2-q24.3 (SPG3), 2p21-p24 (***SPG4***) and 15q11.1 (SPG6). The repeat expansion detection (RED) method has been used to demonstrate expanded CAG repeats in some FSP families that map to ***SPG4***. We analyzed 20 FSP families, including four for which there is evidence for linkage to ***SPG4*** , and found that in most cases the repeat expansion detected by RED is due to non-pathogenic expansions of the chromosome 18q21.1 SEF2-1 or 17q21.3 ERDA1 locus. Polymorphic expansions at SEF2-1 and ERDA1 appear frequent and may confound RED studies in the search for genes causing disorders demonstrating anticipation. In six FSP families, however, CAG repeat expansion was detected in a subset of affected and at-risk individuals that did not result from expansion of the SEF2-1 and ERDA 1 loci. Overall, 11 of 37 (30%) of the FSP patients with a CAG/CTG repeat expansion are unaccounted for by the SEF2-1 and ERDA 1 loci, compared with two of 23 (9%) of the unaffected at-risk individuals and none of 19 controls. In the majority of cases these novel expansions were shorter than those previously reported

L3 ANSWER 8 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS
INC. on STN
DUPLICATE 6
AN 1999:160939 BIOSIS
DN PREV199900160939
TI Quality assessment of whole genome mapping data in the refined familial spastic paraplegia interval on chromosome 14q.
AU Paternotte, Caroline, Rudnicki, Doda, Fizames, Cecile, Davoine, Claire-Sophie; Mavel, Delphine, Durr, Alexandra, Samson, Delphine; Marquette, Catherine, Muselet, Delphine; Vega-Czarny, Nathalie; Drouot,

Nathalie; Voit, Thomas; Fontaine, Bertrand; Gyapay, Gabor; Auburger, Georg; Weissenbach, Jean; Hazan, Jamile [Reprint author]
CS URA CNRS 1922, Genethon, 91000 Evry, France
SO Genome Research, (Nov., 1998) Vol. 8, No. 11, pp. 1216-1227. print. ISSN: 1088-9051
DT Article
LA English
ED Entered STN: 16 Apr 1999
Last Updated on STN: 16 Apr 1999
AB Autosomal dominant familial spastic paraplegia (AD-FSP) is a genetically heterogeneous neurodegenerative disorder characterized by progressive spasticity of the lower limbs. Three loci on chromosome 14q (SPG3), 2p (***SPG4***), and 15q (SPG6) were shown to be responsible for AD-FSP. Analysis of recombination events in three SPG3-linked families allowed us to narrow the critical interval from 9 to 5 cM. An apprx5-Mb YAC contig comprising 32 clones and 90 STSs was built from D14S301 to D14S991, encompassing this region of 14q21. Fifty-six ESTs assigned previously to this region with radiation hybrid (RH) panels Genebridge 4 and G3 were precisely localized on the YAC contig. The 90 STSs positioned on the contig were tested on the TNG RH panel to compare our YAC-based map with an RH map at a high level of resolution. Comparison between our map and the whole genome mapping data on this interval of chromosome 14q is discussed.

L3 ANSWER 9 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN
DUPLICATE 7
AN 1998.347216 BIOSIS
DN PREV199800347216
TI Clinical and genetic analysis of four Swiss families with the pure form of hereditary spastic paraplegia
AU v Fellenberg, J.; Paternotte, C.; Prud'homme, J. F.; Weissenbach, J.; Hazan, J.; Burgunder, J.-M. [Reprint author]
CS Neurogenetische Sprechstunde fuer Erwachsene, Neurologische Poliklinik, Inselspital, CH-3010 Bern, Switzerland
SO Schweizerische Medizinische Wochenschrift, (June 27, 1998) Vol. 128, No. 26, pp. 1043-1050. print.
CODEN: SMWOAS. ISSN: 0036-7672
DT Article
LA German
ED Entered STN: 13 Aug 1998
Last Updated on STN: 13 Aug 1998
AB Hereditary spastic paraplegia (HSP) is a rare neurodegenerative disease of the spinal cord with a progressive gait disorder, associated with other neurological abnormalities in the complicated form. A cluster of families with this disorder in the central part of the country has long been known to Swiss neurologists. In the present report, we describe our clinical and molecular findings in four large families originating from this region and suffering from a pure HSP form. Clinical presentation was similar in the four families. The age of onset varied widely from 2 to 70 years with the appearance of a gait disorder, which slowly progressed to wheelchair confinement after 30-70 years. No other neurological abnormality was found except for impairment of the vibration sense and sphincter abnormalities. In three families an association with markers of the ***SPG4*** locus on chromosome 2 was found. In the fourth, the largest one, no linkage could be found with either ***SPG4***, or with the other two known loci, SPG3 on chromosome 14 and SPG6 on chromosome 15. These data demonstrate the genetic heterogeneity in HSP, even in families from the same region. They also suggest the presence of at least one additional locus for the pure form.

L3 ANSWER 10 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 8
AN 1998.260510 BIOSIS
DN PREV199800260510
TI Autosomal dominant hereditary spastic paraparesis with cognitive loss linked to chromosome 2p
AU Webb, Stewart [Reprint author]; Coleman, David; Byrne, Paula; Parfrey, Nollaig; Burke, Teresa; Hutchinson, Judith; Hutchinson, Michael
CS Dep. Neurol., Southern Gen. Hosp. NHS Trust, 1345 Govan Rd., Glasgow G51 4TF, UK
SO Brain, (April, 1998) Vol. 121, No. 4, pp. 601-609. print
CODEN: BRAIAK. ISSN: 0006-8950.
DT Article
LA English
ED Entered STN: 9 Jun 1998
Last Updated on STN: 9 Jun 1998
AB A family initially considered to have 'pure' autosomal dominant hereditary spastic paraparesis (HSP), was found on neuropsychological testing to have evidence of late onset cognitive impairment. This family showed genetic linkage to the ***SPG4*** locus on chromosome 2p previously reported for pure HSP. Of 56 living members, 44 were examined, 30 of whom were >30 years of age and 12 members were found to be affected with HSP including four asymptomatic cases. One other family member (III-5), aged 62 years, died prior to this study of a 4-year dementing illness. Neuropsychological assessment of 11 affected members and 11 matched unaffected, family controls showed no significant differences between the two groups. However, the neuropsychological test profile in four of 11 affected members tested (mean age 47.2 years) and one of 11 family controls (mean age 41.5 years) showed global cognitive impairment. The pattern of cognitive dysfunction was the same for all five family members identified and was similar to that found in subcortical dementia. The presence of cognitive impairment appeared to be related to age and not the severity of the paraplegia. Both the severity of the paraplegia and the age of onset (21-60 years) varied considerably in this family.

L3 ANSWER 11 OF 23 CAPLUS COPYRIGHT 2003 ACS on STN
AN 1999:155787 CAPLUS
DN 130:350698
TI Linkage of AD HSP and cognitive impairment to chromosome 2p, haplotype and phenotype analysis indicates variable expression and low or delayed penetrance
AU Byrne, Paula C.; Webb, Stewart, McSweeney, Fergus, Burke, Teresa, Hutchinson, Michael, Parfrey, Nollaig A.
CS Departments of Pathology, University College Dublin and St Vincent's Hospital, Dublin, Ire.
SO European Journal of Human Genetics (***1998***), 6(3), 275-282
CODEN: EJHGEU; ISSN: 1018-4813
PB Stockton Press
DT Journal
LA English
AB We report linkage of a family affected with autosomal dominant hereditary spastic paraparesis (HSP) and/or cognitive impairment to the HSP locus on chromosome 2p. To date all families linked to this locus have been affected with 'pure' HSP. The specific pattern of cognitive impairment in this family is characterized primarily by deficits in visuo-spatial functions. We also present genetic studies that indicate variable expression and low or delayed penetrance. We have constructed a haplotype flanked by polymorphic markers D2S400 and D2S2331 that was present in 12 individuals affected with spastic paraparesis. The severity of spasticity varied markedly among these individuals. In addn. four of these individuals (aged 62-70) also had a specific form of cognitive impairment. The disease haplotype was also present in an individual (age 57) who had an identical pattern of cognitive impairment as the only sign of the disease supporting the hypothesis that spastic paraparesis and cognitive impairment are the result of variable expression of a single gene (rather than a co-incidental occurrence). Haplotype reconstruction for all participating family members revealed the presence of this disease haplotype in six individuals who had normal neurol. and neuropsychol. examns. All six are below the maximal age of onset in the family - 60 yr. This is evidence for low or late penetrance of the AD HSP gene in this family. The identification of normal individuals carrying the disease haplotype demonstrates the importance of genetic studies in combination with clin. examn. when counseling at risk family members.
RE.CNT 31 THERE ARE 31 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 12 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN
DUPLICATE 9
AN 1998:165895 BIOSIS
DN PREV199800165895
TI Mapping of a complicated familial spastic paraplegia to locus ***SPG4*** on chromosome 2p.
AU Heinzlef, Olivier [Reprint author]; Paternotte, Caroline; Mahieux, Florence; Prud'homme, Jean-Francois, Dien, Joelle; Madigand, Michel, Pouget, Jean; Weissenbach, Jean, Roullet, Etienne; Hazan, Jamile
CS Serv. Neurol., Hop. Tenon, 4 rue de Chine, 75020 Paris, France
SO Journal of Medical Genetics, (Feb., 1998) Vol. 35, No. 2, pp. 89-93. print.
CODEN: JMDGAE. ISSN: 0022-2593
DT Article
LA English
ED Entered STN: 6 Apr 1998
Last Updated on STN: 6 Apr 1998
AB Autosomal dominant familial spastic paraplegia (AD-FSP) is a degenerative disorder of the central motor system characterised by progressive spasticity of the lower limbs. AD-FSP has been divided into pure and complicated forms. Pure AD-FSP is genetically heterogeneous, three loci have been mapped to chromosomes 14q (SPG3), 2p (***SPG4***), and 15q (SPG6), whereas no loci responsible for complicated forms have been identified to date. Here we report linkage to the ***SPG4*** locus in a three generation family with AD-FSP complicated by dementia and epilepsy. Assuming that both forms of AD-FSP are caused by mutations involving the same FSP gene, analysis of recombination events in this family positions the ***SPG4*** gene within a 0 cM interval flanked by loci D2S2255 and D2S2347.

L3 ANSWER 13 OF 23 EMBASE COPYRIGHT 2003 ELSEVIER INC. ALL RIGHTS RESERVED.
on STN DUPLICATE 10
AN 1999051905 EMBASE
TI Transcript map of the chromosome 2-linked autosomal dominant spastic paraplegia (***SPG4***) critical region and identification of a highly informative STRP [2].
AU Lau E.-L.; Kostrzewa M.; Muller U.
CS U. Muller, Institut fur Humangenetik, Schlangenzahl 14, D-35392 Giessen, Germany. ulrich.mueller@humangenetik.med.uni-giessen.de
SO Neurogenetics, (1998) 2/1 (75-76)
Refs: 3
ISSN: 1364-6745 CODEN: NEROFX
CY Germany

DT Journal, Letter
FS 008 Neurology and Neurosurgery
022 Human Genetics
LA English

L3 ANSWER 14 OF 23 CAPLUS COPYRIGHT 2003 ACS on STN
AN 1997:675710 CAPLUS
DN 128:2571
TI CAG repeat expansion in autosomal dominant pure spastic paraplegia linked to chromosome 2p21-p24
AU Nielsen, Jorgen E ; Koefoed, Pernille; Abell, kathrine; Hasholt, Lis; Eiberg, Hans; Fenger, Kirsten; Niebuhr, Erik; Sorensen, Sven Asger
CS Dep Med Genet., Sect Neurogenet., Panum Inst , Univ Copenhagen, Copenhagen, DK-2200, Den
SO Human Molecular Genetics (***1997***), 6(11), 1811-1816
CODEN: HMGEE5; ISSN: 0964-6906
PB Oxford University Press
DT Journal
LA English
AB CAG repeat expansions have been identified as the disease-causing dynamic mutations in the coding regions of genes in several dominantly inherited neurodegenerative disorders, including spinobulbar muscular atrophy, Huntington's disease, dentatorubral-pallidoluysian atrophy, spinocerebellar ataxia type 1, 2 and 6 and Machado-Joseph disease The CAG repeat expansions are translated to elongated polyglutamine tracts and an increased size of the polyglutamine tract correlates with anticipation, the cardinal feature, seen in all these diseases. Autosomal dominant pure spastic paraplegia (ADPSP) is a degenerative disorder of the central motor system clin characterized by slowly progressive and unremitting spasticity of the legs, hyperreflexia and Babinski's sign Like the established CAG repeat diseases ADPSP is characterized by both inter- and intrafamilial variation and anticipation Using the Repeat Expansion Detection (RED) method, we have analyzed 21 affected individuals from six Danish families with the disease linked to chromosome 2p21-p24 We found that 20 of 21 affected individuals showed CAG repeat expansions vs. two of 21 healthy spouses, demonstrating a strongly statistically significant assocn. between the occurrence of the repeat expansion and the disease (Fisher's test, P < 10-5) suggesting that a CAG repeat expansion is involved presumably as a dynamic mutation in ADPSP linked to chromosome 2p21-p24. The size of the expansion is estd. to be gtoreq 60 CAG repeat copies in the affected individuals. The CAG repeat expansion is very likely translated and expressed as indicated by the detection of a polyglutamine-contg protein in an ADPSP patient
RE.CNT 28 THERE ARE 28 CITED REFERENCES AVAILABLE FOR THIS RECORD
ALL CITATIONS AVAILABLE IN THE RE FORMAT

L3 ANSWER 15 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 11
AN 1997:156805 BIOSIS
DN PREV199799456008
TI Hereditary spastic paraplegia LOD-score considerations for confirmation of linkage in a heterogeneous trait.
AU Dube, Marie-Pierre; Mlodzienski, Melinda A., Kibar, Zoha; Farlow, Martin R., Ebers, George; Harper, Peter, Kolodny, Edwin H , Rouleau, Guy A [Reprint author], Figlewicz, Denise A
CS Montreal General Hosp Research Inst., 1650 Cedar Ave , Room L7-126, Montreal H3G 1A4, PQ, Canada
SO American Journal of Human Genetics, (1997) Vol 60, No 3, pp 625-629.
CODEN: AJHGAG ISSN: 0002-9297
DT Article
LA English
ED Entered STN 15 Apr 1997
Last Updated on STN: 15 Apr 1997
AB *Hereditary spastic paraplegia (HSP)* is a degenerative disorder of the motor system, defined by progressive weakness and spasticity of the lower limbs. HSP may be inherited as an autosomal dominant (AD), autosomal recessive, or an X-linked trait. AD HSP is genetically heterogeneous, and three loci have been identified so far: SPG3 maps to chromosome 14q, ***SPG4*** to 2p, and SPG4a to 15q. We have undertaken linkage analysis with 21 uncomplicated AD families to the three AD HSP loci We report significant linkage for three of our families to the ***SPG4*** locus and exclude several families by multipoint linkage We used linkage information from several different research teams to evaluate the statistical probability of linkage to the ***SPG4*** locus for uncomplicated AD HSP families and established the critical LOD-score value necessary for confirmation of linkage to the ***SPG4*** locus from Bayesian statistics. In addition, we calculated the empirical P-values for the LOD scores obtained with all families with computer simulation methods Power to detect significant linkage, as well as type I error probabilities, were evaluated. This combined analytical approach permitted conclusive linkage analyses on small to medium-size families, under the restrictions of genetic heterogeneity.

L3 ANSWER 16 OF 23 CAPLUS COPYRIGHT 2003 ACS on STN
AN 1997:633119 CAPLUS
DN 127:315330
TI Autosomal dominant spastic paraplegia linked to chromosome 2p: clinical and genetic studies of a large Japanese pedigree
AU Matsuura, Tohru; Sasaki, Hidenao, Wakisaka, Akemi; Hamada, Takeshi; Moriwaka, Fumio, Tashiro, Kunio
CS Department of Neurology, Hokkaido University School of Medicine, Sapporo, Japan
SO Journal of the Neurological Sciences (***1997***), 151(1), 65-70
CODEN: JNSCAG, ISSN: 0022-510X
PB Elsevier
DT Journal
LA English
AB Autosomal dominant spastic paraplegia (ADSP) is a genetically heterogenous disorder. To date, 3 loci of ADSP have been identified on chromosome 2p, 14q, and 15q, but specific gene mutations remain unknown. To det. the genetic background of ADSP in the Japanese, we studied a large 3-generation pedigree, clin. and genetically. Of the 36 individuals clin. examd , 15 were affected The main feature in the affected individuals was a slowly progressive spastic paraplegia, assocd. with upper limb hyperreflexia (58%), redn. of vibration sense (27%) and bladder disturbance (13%) Age at onset ranged from 13 to 50 yr with a mean of 30 3 +- 14 2 (SD). There were 6 parent-child pairs with anticipation and at least 3 others with 'anti-anticipation'. Linkage with 14q and 15q ADSP loci was excluded, and a highly significant lod score was obtained only in the case of the 2p locus (Zmax = 3.53 for D2S400/D2S352, at .theta = 0 00) Our study is the first to confirm the existence of 2p-linked ADSP in the Japanese. There is a significant variety in age at onset and disease severity in these 2p-linked families, but the implication for underlying ADSP mutation is not clear

L3 ANSWER 17 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 12
AN 1997:131919 BIOSIS
DN PREV199799423732
TI Familial spastic paraparesis. Evaluation of locus heterogeneity, anticipation, and haplotype mapping of the ***SPG4*** locus on the short arm of chromosome 2.
AU Raskind, Wendy H [Reprint author]; Pericak-Vance, Margaret A., Lennon, Felicia, Wolff, John; Lipe, Hillary P , Bird, Thomas D
CS Dep Med., Box 357720, Univ. Washington, Seattle, WA 98195-7720, USA
SO American Journal of Medical Genetics, (1997) Vol. 74, No 1, pp 26-36
ISSN: 0148-7299
DT Article
LA English
ED Entered STN: 25 Mar 1997
Last Updated on STN: 25 Mar 1997
AB Familial spastic paraparesis (SPG) is a clinically and genetically heterogeneous group of disorders At least three loci have been implicated in autosomal dominant pure SPG and mutations in either of two loci may cause the X-linked form. Although the penetrance is high for all forms by age 60, there is wide variation in clinical characteristics, including age of onset Two-point and multipoint linkage analyses in nine families provided supportive evidence that the most common form of SPG is linked to chromosome 2 (***SPG4***) Haplotype analysis localized the critical region to a 6 cM interval between D2S392 and D2S367 By haplotype analysis, the disease in at least one family does not appear to be linked to any of the presently known SPG loci, suggesting that there is at least one additional SPG gene. Evaluation of ages of onset in 11 families gave suggestive evidence for anticipation with mean age of onset in parents (41 3 years) being older than mean age of onset in children (26 9 years, P lt 0.005).

L3 ANSWER 18 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
DUPLICATE 13
AN 1997:23107 BIOSIS
DN PREV199799322310
TI Phenotype of autosomal dominant spastic paraplegia linked to chromosome 2
AU Durr, A. [Reprint author]; Davoine, C.-S., Paternotte, C , Von Fellenberg, J ; Cogilnicean, S ; Coutinho, P., Lamy, C ; Bourgeois, S., Prud'homme, J -F ; Penet, C , Mas, J.-L.; Burgunder, J -M., Hazan, J., Weissenbach, J., Brice, A ; Fontaine, B
CS INSERM U289, Hopital de la Salpetriere, 47 Blvd. de l'Hopital, 75651 Paris Cedex 13, France
SO Brain, (1996) Vol 119, No 5, pp. 1487-1496
CODEN: BRAIAK. ISSN: 0006-8950
DT Article
LA English
ED Entered STN. 15 Jan 1997
Last Updated on STN 15 Jan 1997
AB We report the clinical features of 12 families with autosomal dominant spastic paraplegia (ADSP) linked to the ***SPG4*** locus on chromosome 2p, the major locus for this disorder that accounts for apprx 40% of the families. Among 93 gene carriers, 32 (34%) were unaware of symptoms but were clinically affected Haplotype reconstruction showed that 90% of the asymptomatic gene carriers presented increased reflexes and/or extensor plantar responses independent of age at examination. The mean age at onset was 29 years, ranging from 1 to 63 years. Intra- as well as inter-familial variability of age at onset was important, but did not result from anticipation. Phenotype-genotype correlations and comparison with SPG3 and SPG5 families indicated that despite the variability of age at onset, ***SPG4*** is a single genetic entity but no clinical features distinguish individual ***SPG4*** patients from those with SPG3 or SPG5 mutations.

L3 ANSWER 19 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC. on STN
AN 1996:561943 BIOSIS

DN PREV199699284299
TI YAC contig map of the candidate region for familial spastic paraplegia (***SPG4***) on chromosome 2p21 fwdarw p14
AU Krols, L. [Reprint author], Michalik, A. [Reprint author], De Jonghe, P. [Reprint author], Martin, J.-J., Van Broeckhoven, C.
CS Lab. Neurogenet., Dep. Biochem., Univ. Antwerp, Antwerpen, Belgium
SO Cytogenetics and Cell Genetics, (1996) Vol. 73, No. 4, pp. 271. Meeting Info.: Fourth International Workshop on Human Chromosome 2 Mapping. London, England, UK. April 10, 1996. CODEN: CGCGBR. ISSN: 0301-0171.
DT Conference; (Meeting). Conference, Abstract; (Meeting Abstract)
LA English
ED Entered STN: 13 Dec 1996. Last Updated on STN: 13 Dec 1996

L3 ANSWER 20 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN. DUPLICATE 14
AN 1997:42891 BIOSIS
DN PREV199799334879
TI Pure familial spastic paraplegia: Clinical and genetic analysis of nine Belgian pedigrees
AU De Jonghe, Peter, Krols, Luc, Michalik, Andrej, Hazan, Jamiel; Smeyrs, Gisele, Lofgren, Ann; Weissenbach, Jean, Martin, Jean-Jacques, Van Broeckhoven, Christine [Reprint author]
CS Lab. Neurogenetics, Univ. Antwerp, Dep. Biochem., Universiteitsplain 1, B-2610 Antwerpen, Belgium
SO European Journal of Human Genetics, (1996) Vol. 4, No. 5, pp. 260-266. ISSN: 1018-4813.
DT Article
LA English
ED Entered STN: 28 Jan 1997. Last Updated on STN: 28 Jan 1997
AB We ascertained 9 multigeneration Belgian families with pure dominant spastic paraplegia (SPG) for clinical and genetic studies. Linkage was examined using simple tandem repeat (STR) markers located near the 5 loci for familial SPG on chromosomes Xq28 (SPG1), Xq21.3-q22 (SPG2), 2p21-p24 (***SPG4***), 14q12-q23 (SPG3) and 15q11.1 (SPG6). Positive linkage results were obtained only for markers at the ***SPG4*** locus mapping the ***SPG4*** gene between D2S400 and D2S367, a region of 4 cM. In order to facilitate the positional cloning of the ***SPG4*** gene, we constructed a contiguous YAC map covering the ***SPG4*** candidate region. Our physical mapping data indicate that the ***SPG4*** gene resides within maximal 5 Mb.

L3 ANSWER 21 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN
AN 1996:355674 BIOSIS
DN PREV199699078030
TI YAC contig map of the candidate region for familial spastic paraplegia (***SPG4***) on chromosome 2p14-p21
AU Krols, Luc [Reprint author]; Michalik, A. [Reprint author], De Jonghe, P. [Reprint author]; Martin, J.-J., Van Broeckhoven, C. [Reprint author]
CS Lab. Neurogenet., Dep. Biochem., Born-Bunge Found., Univ. Antwerp, Antwerpen, Belgium
SO European Journal of Human Genetics, (1996) Vol. 4, No. SUPPL. 1, pp. 100. Meeting Info.: 28th Annual Meeting of the European Society of Human Genetics. London, England, UK. April 11-13, 1996. ISSN: 1018-4813
DT Conference, (Meeting). Conference; Abstract; (Meeting Abstract). Conference, (Meeting Poster)
LA English
ED Entered STN: 5 Aug 1996. Last Updated on STN: 5 Aug 1996

L3 ANSWER 22 OF 23 CAPLUS COPYRIGHT 2003 ACS on STN
AN 1988:624758 CAPLUS
DN 109:224758
TI Contractile system of spasmoneme
AU Ochiai, Tsutomu; Asai, Hiroshi
CS Coll. Sci. Tech., Waseda Univ., Tokyo, Japan
SO Seitai no Kagaku (***1988***), 39(2), 89-91. CODEN: SEKAA6, ISSN: 0370-9531
DT Journal, General Review
LA Japanese
AB A review, with 13 refs., on mechanisms and components (proteins, such as ***spastin***) of the contractile system of protozoan spasmoneme.

L3 ANSWER 23 OF 23 BIOSIS COPYRIGHT 2003 BIOLOGICAL ABSTRACTS INC on STN. DUPLICATE 15
AN 1983:184467 BIOSIS
DN PREV198375034467; BA75:34467
TI EXTRACTION AND SOME PROPERTIES OF THE PROTEINS ***SPASTIN*** B FROM THE SPASMONEME OF CARCHESIUM-POLYPINUM
AU YAMADA K [Reprint author]; ASAI H
CS DEP PHYSICS, SCH SCIENCE ENGINEERING, WASEDA UNIV, SHINJUKU-KU, TOKYO 160
SO Journal of Biochemistry (Tokyo), (1982) Vol. 91, No. 4, pp. 1187-1196. CODEN: JOBIAO. ISSN: 0021-924X
DT Article
FS BA
LA ENGLISH
AB Proteins of the contractile spasmoneme from C. polypinum were extracted in 2% SDS [sodium dodecyl sulfate] 30% acetic acid, or 8 M urea. The proteins extracted in SDS had a wide MW distribution when examined by SDS-polyacrylamide gel electrophoresis. The proteins extracted in urea and acetic acid had 3 major peaks with MW of about 16,000, 18,000 and 22,000. Most of these proteins were soluble even in the absence of urea and were monomeric, since the sedimentation coefficient, S20,w, measured by analytical ultracentrifugation was 2.0S. The electrophoretic mobility of the proteins extracted in urea or in acetic acid was examined on alkaline gels. In the presence of free Ca2+, the mobility was significantly reduced compared with that in the absence of free Ca2+. These Ca-binding proteins were heat-stable and could not interact with troponin I. The implications of these proteins and others in relation to the contractility of the spasmoneme in Carchesium stalk are discussed.

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